

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: April 24, 2014

Subcommittee Members Attending:

Barbara Burton, Lurie Children's Hospital – Chair
Darrel Waggoner, Lainie Friedman-Ross - University of Chicago
Zohra Shad, Rich Dineen - University of Illinois Chicago
Brook Croke, Jennifer Burton - University of Illinois Chicago at Peoria
Tess Rhodes - Division of Specialized Care for Children

IDPH Staff:

Khaja Basheeruddin, Rong Shao, Claudia Nash, Shannon Harrison, Nikki Woolverton, Heather Shryock

Background

The meeting was called to order at 2:05 PM.

IDPH Laboratory Status Report

Khaja Basheeruddin reported 15,000 samples have been analyzed for lysosomal storage disorders (LSDs) using IDPH tandem mass spectrometry. Statewide samples are being screened, and positives are de-identified and sent to Perkin Elmer for validation. Twenty five of the 15,000 samples have been sent to Perkin Elmer Genetics Laboratory for validation of abnormal screening results. In addition, 13 samples have been sent to the New York State Health Department Laboratory for Krabbe testing. IDPH laboratory has reported a total of eight abnormal newborn screening specimens for LSDs to the NBS follow up program; three Pompe, three MPS I, one Niemann-Pick and one Gaucher. IDPH laboratory is expecting to start pilot testing with Northwestern Memorial Hospital on May 19, 2014, and LSD results for Northwestern will be issued by IDPH. IDPH lab will continue to test statewide samples for LSDs and send any abnormal results to Perkin Elmer Genetics for confirmatory testing. For specimens from all hospitals except Northwestern, a Perkin Elmer Genetics report will be used when the test is abnormal. The IDPH report will not reflect LSD testing for these babies. IDPH laboratory stated there are still some database changes that need to be made by Perkin Elmer before statewide LSD screening can be documented in the IDPH database for the July 1, 2014 start date.

Several concerns were raised by Dr. Burton, Dr. Waggoner and Dr. Ross regarding statewide screening for LSDs by IDPH laboratory in order to validate their testing. Concerns expressed were:

- NBS database is not ready to accept LSD results; therefore, tracking of these abnormal NBS will be difficult and data will not be able to be accurately retrieved
- Hospitals and physicians do not know if newborns under their care had LSD screening or not from the current report that is being sent to hospitals
- LSD results are being sent to physicians using a Perkin Elmer report; this is confusing to physicians
- Some Advisory Committee members indicated they were not aware this would be the process used by the IDPH laboratory for validation of LSD screening
- IDPH laboratory is determining which samples are being tested using a randomized approach which was stated by some to be unethical and irresponsible
- Hospitals, physicians and medical specialists were not currently aware of this validation process which contradicts an earlier letter sent out by the IDPH laboratory

Khaja Basheeruddin stated Dr. Dizikes could not attend today's meeting due to an APHL meeting conflict but stated he would inform Dr. Dizikes of the committee's concerns. The committee asked to Claudia Nash to schedule another meeting with Dr. Dizikes in the near future to address these concerns. Claudia Nash will send out possible meeting dates to the group and schedule another meeting within the next two weeks.

Meeting adjourned 2:45 p.m.